Lung Diseases: Molecular Basis Comparison

Disease	Molecular Defect	Mechanism	Clinical Impact
RDS (Premature)	Insufficient surfactant production	Inadequate DPPC and SP-B in alveoli causes increased surface tension	Lung collapse in pre-term infants; 100% incidence at 24-26 weeks gestation
Hereditary RDS Chromosome 2	SP-B gene mutation (2 bp insertion at codon 121)	Frame-shift mutation causes complete absence of SP-B protein	Progressive respiratory failure unresponsive to treatment; fatal without lung transplant
Emphysema Chromosome14	α1-AT deficiency (GAG→AAG mutation at codon 342)	Glu→Lys substitution prevents protein folding; elastase degrades alveolar elastin unchecked	Irreversible alveolar wall destruction; smoking oxidizes Met-358 worsening condition
Cystic Fibrosis Chromosome 7	CFTR gene mutation (3-base deletion at codon 508)	Defective Cl- pump causes mucus thickening via water retention	Thick mucus accumulation in lungs; fatal by age 5 without treatment
Immotile Cilia Syndrome	Dynein arm, radial spoke, or microtubule defects	Defective ciliary structure (9+2 arrangement) prevents mucociliary clearance	Chronic respiratory infections; permanent lung damage; may require transplantation